

Newborn Baby Metabolic Screening

Age(s) of child

2-4 days

Purpose

In some genetic disorders early diagnosis, sometimes even before symptoms begin, can lead to treatment that improves outcomes. In New Zealand screening of all newborn babies is undertaken after the second day of life for certain genetic diseases. These conditions include phenylketonuria (PKU), maple syrup urine disease (MSUD), biotinidase deficiency, cystic fibrosis (CF), congenital hypothyroidism (CH), galactosaemia, and congenital adrenal hyperplasia (CAH).

Personnel

The Lead Maternity Carer (LMC - obstetrician, midwife or general practitioner) is responsible for ensuring this screening procedure is undertaken.

Recommended procedure

- Providers must have current competency in collection of blood samples from newborn babies and practice correct disposal of blood collection equipment
- Follow the National Committee Clinical Laboratory Standards.

Educational preparation needs to include:

- informed consent processes
- blood collection onto filter paper
- correct disposal of blood collection equipment.

Procedure

Gain informed consent of parent. The LMC needs to ensure that consent is available if blood collected by someone other than them.

Samples are to be collected after 48 hours of protein feeding in a normal healthy baby and preferably as soon as possible after the 48 hours. If the baby is premature or ill or not feeding at 48 hours, a sample should be taken, and a second sample after 48 hours of protein feeding.

The sample must include the name and contact telephone number of the LMC. The telephone number must be one that will give immediate access to the LMC as urgent contact may be required for positive test results. Telephone numbers that have an answer phone most of the time are not acceptable.

Blood collection

1. Do not touch the specimen collection paper (circles).
2. Fill out all requested information on both sections of the test card.
3. Ensure caregiver has seen the factsheet *Your Newborn Baby's Blood Test*.
4. Warm foot if necessary.
5. Sterilise the puncture site (as in diagram) and wipe dry.
6. Make puncture with lancet (tip shorter than 2.4mm).
7. Gently apply collection paper to large drop of blood. Allow blood to soak through completely. If blood does not fill the circle another drop may be applied immediately. Collect from one side of the paper only and check the circle is completely filled on both sides.
8. Repeat step 7 to fill the other 3 circles.
9. Allow to dry in a cool place (2-4 hours).



The quality of the screening is critically dependent on sample quality. If the blood is not flowing freely and the blood becomes layered on the paper too much blood will be put into the tests and false positive results may occur. If the blood is not soaked right through the paper from one side not enough blood will be put in the tests and false negative (missed cases) results may occur.

Cardboard racks suitable for drying samples can be obtained from the National Testing Centre. In hot weather these should be put in the shade eg, into a chilly-bin out of the sun.

Mail cards DAILY to:

National Testing Centre
P O Box 872
Auckland

ph (09) 307 4949 x6570
fax (09) 307 4936

or

Courier delivery to:

Specimen Reception
LabPlus
Building 31 (Gate 4)
Auckland Hospital
Grafton Rd
Auckland

Referral pathway

Requests for follow-up will be made when an initial sample is unsuitable for testing or when there are abnormal test results. All positive tests for cystic fibrosis (CF) and biotinidase deficiency, and slightly abnormal results for the other screening tests will be communicated by letter. Notification of very abnormal results for maple syrup urine disease (MSUD), phenylketonuria (PKU), galactosaemia, congenital hypothyroidism (CH) and congenital adrenal hyperplasia (CAH) will be made by telephone.

MSUD, galactosemia and CAH are life-threatening and it is important the contact numbers supplied allow immediate contact with the LMC. The telephone call from the National Testing Centre will include a recommendation for paediatrician referral and for diagnostic tests.

Resources

Test cards and parent information sheets are available at no charge from the National Testing Centre.

Standards

Parent information sheets will provide appropriate information to ensure parents can make an informed decision and consent for this procedure.

The US National Committee on Clinical Laboratory Standards (NCCLS) has a standard for newborn baby blood collection onto filter paper.

There are no standards for the screening testing or follow-up although these are currently being developed by the Joint Newborn Screening Committee of the Human Genetics Society of Australasia and the Royal Australian College of Physicians: see <http://www.hgsa.com.au/> under HGSA Committees.

Documentation

It is not possible at this time for the National Testing Centre to compare test records with either birth information or health registration information in order to determine or ensure coverage by the screening programme. Local documentation will need to be kept to ensure all infants are tested.

The Centre keeps a record of follow-ups requested and received. When test results are very abnormal the Centre will ensure follow-up has occurred. If follow-up has not occurred within one month the Centre will send a reminder letter. Resources are insufficient to ensure complete follow-up.

Every month the Centre will send a list to LMCs of the babies who have been tested in the preceding month. This is a list only and does not have results. It is sent so LMCs can ensure all the infants in their care have had a test.

Rationale

Rationale, including a favourable cost-benefit analysis of newborn screening tests, is reflected in the recommendations of the Joint Committee referred to above.

These are:

- Screening is unequivocally recommended for phenylketonuria and primary congenital hypothyroidism.
- There are arguments in favour of screening for cystic fibrosis, congenital adrenal hyperplasia and galactosaemia.
- Screening tests for maple syrup urine disease and biotinidase deficiency exist, but evidence that the advantages of early diagnosis outweigh costs is at present insufficient to recommend their routine use.
- Some aspects of biochemical genetics and at-risk metabolic screening services are not presently provided in New Zealand. This is a factor in the decision to do some screening in New Zealand that is not recommended for Australia. The newborn screening tests provided in New Zealand are reviewed at the time of contract negotiations. Where screening does not benefit the screened infants (eg, they are detected by the metabolic service before the screen test result is available) the screening will be discontinued.